19 Pancreatic sufficiency and insufficiency in cystic fibrosis newborn screening (CF NBS)

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Aim: Evaluation of patients with pancreatic sufficiency (PS) and insufficiency (PI) diagnosed in CF NBS.
Methods: Groups of 17 PS and PI patients diagnosed in CF NBS with the similar CFTR mutations and confirmed elastaze-1 status were analyzed. Patients with Meconium ileus were excluded. Birth weight and length, age at diagnosis, IRT, sweat test values, evaluation of SD body weight and height at the end of the 1st year of life as well as *Pseudomonas aeruginosa* (PA) infection were collected.
Results: From Sept. 2006 to the end of Dec. 2010 CF was confirmed in 84 newborns screened. In PS group were 41% females and 53% respectively. Both groups consisted of 10 F508del heterozygotes and 7 non-F508del heterozygotes. Average age of diagnosis in PS group was similar to PI patients (55 v 45 days). The mean birth weight (3697.35±472.45 v 3122.12±368.52; $p < 0.001$) and length (55.82±2.83 v 53.76±2.49; $p = 0.031$) were significantly higher in PS patients. The mean IRT for the group PS was significantly lower (455.07±342.88 v 676.72±353.22; $p = 0.036$). At the end of 1st year of life, mean SD body weight and IWH% was in the normal range without differences between group. However, SD height (0.36±0.81 v –0.26±0.89; $p = 0.033$ were significantly reduced in PI patients. Iontophoresis sweat test was significantly lower in PS group (54.48±20.55 v 81.31±22.77; $p = 0.001$). 3 patients in both groups had infection with PA in assessed period.
Conclusion: CF newborns with PS were characterized by lower sweat test and IRT results but higher birth weight and length than PI children. Although there were no differences in weight at the end of 1st year of life between the groups, children with PI were taller.

20 Cystic fibrosis newborn screening in Italy: survey for assessment of technical-scientific and organizational issues

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Background: In Italy CF newborn screening (CFNBS) is mandatory since 1992 and there are different regional approaches.
Aim: The aim is to describe the state of the art of CFNBS programs in Italy.
Material and Methods: This is a survey, descriptive study. Data relative to technical, organizational and clinical CFNBS aspects were collected by 15/15 labs involved in CFNBS and 16 CF Care Centers. Data were noted by 3 research assistants trained in audit visits; individual data of newborn screened CF infants were also evaluated according 30 process and outcome indicators.
Results: Although mandatory, only 15/20 regions in Italy implemented CFNBS using 5 different protocols 7 = IRT/DNA/IRT; 4 IRT/IRT; 1 IRT; 1 IRT/meconium lactase; 1 IRT/DNA+ meconium lactase.
In the year 2009 445,118 newborn (78% of Italian population) were screened with incidence of 1:5000 (1:2400–1.12000). This study included 124 newborn screened CF infants, born in 2009 and diagnosed within the first year of life. 19/124 had meconium ileus; 104 were classic CF and 19 were atypical CF.
Median age at diagnosis was 56 days (1rts and 3rst quartile 30 and 67 days)
Median age at first visit at CF Center was 67 days. Out of 124 screened infants 12.7% presented wasting and 15% presented stunting at diagnosis, 12.7% had wasting and 10.7% had stunting at age of 1 years; 6.7% were chronically colonized with *Pseudomonas aeruginosa* within first year of life. Other major critical issues concern communication and organizational area.
Conclusion: There are different regional approaches to CF newborn screening (CFNBS) and within the same region where multiple CFNBS centres coexist: these data may guide the implementation of CNBS in Italy.

21 Diagnostic and counselling dilemmas in newborn screening for cystic fibrosis related to the detection of atypical mutations

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Objectives: Newborn screening (NBS) for CF has been implemented as a nationwide IRT/DNA/IRT scheme in the Czech Republic since October 2009. DNA testing is associated with inherent drawbacks such as detection of infants with atypical mutations resulting in variable phenotypes.
We evaluated a clinical status in individuals (from NBS and non-NBS group) carrying either R117H or D1152H allele in *trans* with another CF-causing mutation and utilized these data for CF NBS and genetic counselling.
Methods: The Czech CF registry and an "in house" clinical-genetic database were used.
Conclusions: Of 13 individuals with CF-causing mutations/R117H on a 7T background, 6 symptomatic adults (1 suffering from respiratory symptoms (RS), 1 from pancreatitis, 4 with azoospermia) and 1 child with RS were reported before implementation of CF NBS. 1 adult with unknown clinical status was identified due to cascade screening. 5 infants were identified in CF NBS without having CF symptoms. There has been no case with CF-causing mutation/R117H on a 5T background yet. Of 5 individuals with F508del/D1152H genotype, 2 children and 2 adults suffer from RS and 1 adult from pancreatitis and azoospermia. These cases were reported before implementation of CF NBS and none from CF NBS, so far.
Although phenotype in individuals with CF-causing mutation/R117H-7T genotype is usually mild (mainly CBAVD), we follow consensus guidelines and monitor infants on a long-term basis. Albeit limited knowledge exists on phenotypes associated with D1152H, this mutation is considered to be CF-causing mutation and long-term follow up in a CF specialist is essential.
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22 Screening for cystic fibrosis related complications in a paediatric population

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Objectives: To assess if our tertiary paediatric cystic fibrosis (CF) centre is adhering to best practice guidelines for screening for CF related complications as outlined by the CF Foundation (CFF).
Methods: Retrospective review of annual screen reports of the 90 CF patients attending our paediatric tertiary centre in 2011.
Results: All patients attending our centre ($n = 90$) had liver function tests performed in 2011 and all children $\geqslant$5 years old (60/90, 67%) had a liver ultrasound performed. All patients (6/90, 7%) whose liver ultrasound was concerning for cirrhosis were referred to a paediatric hepatologist. Annual vitamin D levels were performed on all patients and dexa scans were performed on all patients $\geqslant$10 years old with BMI $\leqslant$9th centile or those who had significant doses of oral steroids in the previous year (11/90, 12%).
Annual fasting blood glucose levels were performed on all patients <8 years old and an oral glucose tolerance test was performed on all children $\geqslant$8 years old. HaemoglobinA1c was measured in 84/90 (93%) patients in 2011. Renal function was checked on all patients both at annual screen and on each hospital admission, weekly if treated with aminoglycosides.
Our dedicated CF psychologist attends CF clinic weekly and in addition reviews patients at her own clinic separately if needed. Our CF psychologist reviewed 59/90 (66%) patients in 2011, of which 36/59 (61%) had needle phobia and 33/59 (56%) had feeding related issues as reasons for review.
Conclusions: Our tertiary paediatric CF service compares favourably with CFF guidelines and figures. Our team does however lack a dedicated specialist CF pharmacist. This need is being addressed.